t(2;22)(p23;q11.2)

Jean-Loup Huret

Genetics, Dept Medical Information, UMR 8125 CNRS, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France (JLH)

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**Disease**

Translocations involving 2p23 are found in more than half cases of anaplastic large cell lymphoma (ALCL), a high grade non Hodgkin lymphoma (NHL). They involve ALK, and are therefore called ALK+ ALCL. The most frequent ALK+ ALCL being the the t(2;5)(p23;q35) with NPM1 -ALK fusion protein, which localises both in the cytoplasm and in the nucleus. The t(2;22)(p23;q11.2) is very rare, and, like other t(2;Var) involving various partners and ALK, the fusion protein has a cytoplasmic localization; they are therefore called "cytoplasm only" ALK+ ALCL. However, in case of a t(2;22) the localization is restricted to granules (vesicles) in the cytoplasm.

**Phenotype/cell stem origin**

CD30+; ALK+.

**Clinics**

Found in 1 case (a 3 yr old girl in complete remission 1yr after end of treatment), perhaps 2 (a 52 yr old man).

**Genes involved and proteins**

**ALK**

Location 2p23

Protein 1620 amino acids; 177 kDa; glycoprotein (200 kDa mature protein) ; membrane associated tyrosine kinase receptor.

**CLTCL1**

Location 22q11.2

Protein 1640 amino acids, 187 kDa; component of the coat of vesicles originated from the plasma membrane or the golgi.

**Result of the chromosomal anomaly**

**Hybrid gene**

Description 5' CLTCL1 - 3' ALK.

**Fusion protein**

Description 2197 amino acids, 248-250 kDa; 1634 (nearly all the CLTCL1 protein) N-term amino acids from CLTCL1, fused to the 562 C-term amino acids from ALK (i.e. the entire cytoplasmic portion of ALK with the tyrosine kinase domain).

**Expression / Localisation**

Cytoplasmic localization restricted to granules.

**Oncogenesis**

Constitutive autophosphorylation.

**References**


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